PROFILE

Ciara

LIVING WITH Koolen De Vries Syndrome

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"Ciara is a character and is pure joy. Her laugh lights up any room."

Ciara was 6.14lbs and 20 inchs long at birth. Nothing suggested prenatally what was to come. Ciara passed all prenatal scans with flying colours.

She is our youngest child. Ciara faced many challenges. Ciara was NG fed for a time as a newborn as she was unable to suck the bottle. She was the biggest baby in the Neonatal ICU. She was transferred from the Rotunda to Temple St for further investigations.

Ciara has not met development milestones, with delayed speech, hip issues, eye issues, etc. The list goes on and on. She has seen several specialists such as Cardiologist, Ophthalmologist. She is currently receiving Occupational Therapy, Speech Therapy and Physical Therapy.

As time passed and Ciara's milestones were delayed, we needed answers: Why was Ciara delayed? Why did she look different to her big sister Siobhan?

Ciara's medical team and specialist had no answers. She underwent a number of tests - FISH test, microarray, MRI. Each and every test came back normal. Her blood work for other syndromes also came back negative. Still we kept pressing Ciara's medical team for answers.

Finally in June 2020 after a 6 month wait on the Whole Genome Sequencing we had a result. Ciara was diagnosed with a mutation on KANSL1 in C17q.21.31. (Koolen De Vries Syndrome). We finally had answers and were also scared and confused about what this meant for Ciara's future. We googled and researched everything we could about KDVS. We joined the Facebook groups and have connected with other parents online.

Although KDVS is pretty rare and Ciara has the rarer type, we're still not certain about what the future holds for Ciara. We are relieved though to finally have answers. Ciara is being monitored closely by her medical team and continues to improve.

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